

# Python Package `evo_tri` Documentation

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The `evo_tri` package contains various functions for implementing the evolutionary triangulation algorithm. These functions carry the user through data setup and analysis from downloading or loading files from HapMap or local sources, to parsing files and calculating pairwise *F*<sub>st</sub>s, to running evolutionary triangulation algorithm, to finding genes in vicinity of SNPs found by evolutionary triangulation algorithm, to graphing number of SNPs and genes found over ranges of *F*<sub>st</sub> cutoffs.

This package requires numpy (and matplotlib for `hit_plotter2D()` and `hitplotter3D()` )

The evolutionary triangulation algorithm finds pairwise *F*<sub>st</sub>s for shared SNPs between each pairing of three different populations, filters these SNPs for each population pair based on user-selected *F*<sub>st</sub>s cutoffs (for example, SNPs with *F*<sub>st</sub> values  $\geq .45$ ), and then finds the overlapping set of these filtered SNPs between the three populations of interest. Further analysis can be done once this set of overlapping SNPs has been found: genes in the vicinity of these SNPs can be found, and the number of SNPs found with a certain cutoff can be compared to a range of other *F*<sub>st</sub> cutoffs.

First, population data must be loaded and set up. The `evo_tri` package contains functions that allow the user to download relevant HapMap allele frequency files, use predownloaded HapMap allele frequency files, or load pre-processed custom population data or precalculated *F*<sub>st</sub> values.

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```
evo_tri.evo_hapmapDLer(phase='2009-01_phaseIII',  
pops=['CEU','YRI','GIH'], chrs='autosome,X',  
file_folder="current")
```

`evo_hapmapDLer`: a function for externally downloading hapmap allele\_freq files for an arbitrary number of populations

parameters:

- phase: String that specifies hapmap phase, e.g. '2009-01\_phaseIII'
- pops: list of populations as 3 letter hapmap abbreviations, e.g. 'CEU', or 'YRI'
- chrs: Enter string of desired chromosomes (1-22,X,Y,M) to be downloaded for all populations separated by commas, no spaces.  
Use "autosome" to select all autosomal chromosomes (e.g. "autosome,X,Y"). Use colons to indicate ranges (e.g. "1,4:10,15,Y")
- file\_folder: output folder pathname in which to dump downloaded files. Default to current directory

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```
evo_tri.evo_text2numpy(pop1="CEU", pop2="YRI", pop3="GIH",
customFst=False, no_popdata=False,
pop1filename='pop1textdata.txt',
pop2filename='pop2textdata.txt',
pop3filename='pop3textdata.txt',
pop12filename='pop12textdata.txt',
pop13filename='pop13textdata.txt',
pop23filename='pop23textdata.txt',
file_folder="current", out_compressed=False,
pop_out="pop_archive1", FST_out="FST_archive1")
```

evo\_text2numpy: converts custom data in text format into a .npz (numpy archive) file, for efficient processing

parameters:

```
pop1,pop2,pop3: hapmap population 3 letter abbrev, or custom population
abbreviation.
customFst: True if custom FST data will be provided
no_popdata: if True, no population data file will be created
pop[1-3]filenames: text file name strings for each population to be combined in
numpy array archive. Rows of text file should be each SNP, columns
should be rs number, chromosome number, snp position, allele frequency, and
sample size, in that order, seperated by white space. If certain data not
required for calculation (for example, if custom FST values will be provided),
fill in place holder column with -1's.

SNPs given do not need to be completely overlapping.
pop[12-13-23]FSTfilenames: text file name strings for each population to be
combined in numpy array archive. Rows should be each SNP, columns should be
rs number and associated Fst, seperated by white space. SNPs given do not
need to be completely overlapping.
file_folder: input and output folder pathname.
out_compressed: output file will be compressed
pop_out: file name (without .npz extension) of population raw data
FST_out: file name (without .npz extension) of population FST data
```

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```
evo_tri.evo_tri_data(datasource=2,
data3customFst=False, no_popdata=False,
custom_popdata='pop_archive1.npz',
custom_FSTdata='FST_archive1.npz',
pop1="CEU", pop2="YRI", pop3="GIH", chrs="autosome,X",
unbiasedFst=True,
phase='2009-01_phaseIII', pop_out="pop_data1",
FST_out="fst_data1", out_compressed=False,
file_folder="current")
```

evo\_tri\_data: accesses and sets up data, calculates pairwise Fst values for overlapping SNPs. Saves a dictionary of 2d numpy arrays for each population combo with columns as rs numbers and corresponding Fst for each SNP common to all three populations. Also optionally saves a dictionary of 2d numpy arrays for each population with columns as rs number, chromosome number (X->23,Y->24,M->25), snp position, allele frequency,

and sample size, in that order, for each SNP common to all three populations. See parameters for details.

parameters:

datasource: 1 is hapmap online, 2 is local hapmap txt allele\_freq text files, 3 is local custom data (see below for required format), with or without Fst and gene location data.

data3customFst: Only relevant if data source 3 is selected. If True, will use custom calculated Fst values, see below. Else, will calculate Fst from given data.

no\_popdata: Only relevant if data source 3 is selected and custom Fst will be used.

If true, only SNP/precalculated Fst is taken, allowing for faster calculation. Genefinding will not be possible if this option is used.  
custom\_popdata, custom\_FSTdata: Only relevant if data source 3 is selected. File names for preprocessed custom data in .npz files. To convert text data files to .npz, see evo\_text2numpy(). custom\_popdata file should be .npz dictionary of 3 numpy matrices of floats, one for each population, keyed with '[population abbreviation]'. rows should be SNPs, columns should be rs number, chromosome number, snp position, allele frequency, and sample size, in that order. Chromosomes X,Y and mitochondrial (M) should be coded as 23, 24 and 25 respectively. If certain data are not required for calculation (for example, if custom FST values will be provided), fill in place holder column with -1's. SNPs given do not need to be completely overlapping. If it is being used, custom\_FSTdata file should be .npz dictionary of 3 numpy matrices of floats, one for each population combo (1-2, 1-3, 2-3), keyed with '[population 1 abbreviation] + '[population 2 abbreviation]'. Rows should be each SNP, columns should be rs number and associated Fst, SNPs given do not need to be completely overlapping.

Data required:

If custom Fst values are provided, only Fsts and matching rs-number file are required.

If unbiased Fst will be calculated, at least rs-numbers, allele frequencies, and sample sizes for each population are required.

If uncorrected Fst will be calculated, only rs numbers and allele frequencies are required.

If genefinding will be used, chromosome and snp location data for each population is required.

Each .txt file should be a column of numerical values corresponding to the order of rs numbers for that population provided.

pop1,pop2,pop3: hapmap population 3 letter abbrev, or custom population abbreviation.

chrs: Only relevant with datasource 1 and datasource 2. Enter string of desired chromosomes (1-22,X,Y,M) separated by commas, no spaces. Use "autosome" to select all autosomal chromosomes (e.g. "autosome,X,Y"). Use colons to indicate ranges (e.g. "1,4:10,15,Y")

phase: only relevant if hapmap files will be downloaded (i.e. datasource 1). String that specifies hapmap phase, e.g. '2009-01\_phaseIII'

pop\_out, FST\_out: string with output file name (.npz extension not necessary). For raw population data and calculated Fst data respectively.

out\_compressed: if true, output files are compressed

file\_folder: directory with files, for datasource 2 and 3; directory for storing downloaded file database for datasource 1. Defaults to current directory.

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Once the data has been properly set up and Fst values have been calculated (or provided), the evolutionary triangulation algorithm may be run.

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```
evo_tri.evo_triangler(fst_file='fst_data1.npz',
pops=['CEU','YRI','GIH'], pop12lim=">=.45",
ptile_cutoff=False, pop13lim=">=.45", pop23lim="<=.05",
snps2screen=True, snps2txt=False,
snps_filename="evotri_snps",
file_folder="current")
```

`evo_triangler()`: implements evolutionary triangulation algorithm, saving SNPs found to .npy and (optionally) text files.

parameters:

- `fst_file`: name of dictionary containing Fst data for pairings of three populations (such as `Fst_out` from `evo_tri_data()` )
- `pops`: list of population abbreviations for population 1, population 2, and population 3, in that order
- `ptile_cutoff`: if True, will consider cutoffs entered below as percentiles (in decimal form, e.g. 95% = .95) rather than absolute cutoffs.
- `pop12lim, pop13lim, pop23lim`: Fst threshold (string with operator [`>`, `>`, `>=`, `<=`] followed by Fst between 0 and 1) for pop1-pop2 Fsts, pop1-pop3 Fsts, and pop2-pop3 Fsts respectively
- `snps2screen`: true prints snps found to screen
- `snps2txt`: true prints snps to txt file
- `snps_filename`: name for snps txt file and .npy file
- `file_folder`: directory from which to load and save files. Defaults to current Directory.

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Once the evolutionary triangulation algorithm has been run, further analysis may be performed using the following functions.

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```
evo_tri.genefind_local(snplist='evotri_snps.npy',
custom_loc=False, loc_data='pop_data1', pop='first',
bp_range=100000, custom_gene=False,
custom_genefile='custom_genes.txt',
genes2txt=False, genes2screen=True, genes_filename='genes1',
display_chr=False, file_folder="current")
```

`genefind_local`: finds genes in regions of overlapping SNPs found (within specified range) using local gene location data

parameters:

- `snplist`: .npy file storing array of overlapping SNPs

custom\_loc: if True, uses .npy file storing 3 column 2d numpy array, with rs numbers, corresponding chromosomes, and corresponding locations, in that order, separated by white space. Allows use of custom location file, not generated by evo\_tri\_data.

loc\_data: string with .npy or .npz file name (without extension), with file containing SNP location data. If custom\_loc==True, provide 3 column file (see above). Otherwise, use .npz file in format of pop\_out from evo\_tri\_data().

pop: only relevant if custom\_loc is false. Specify population in loc\_data from which to take SNP locations. If population is not given or not found, population data will be taken from first population in loc\_data.

bp\_range: integer specifying how many bases from a SNP a gene must be to be considered a hit.

custom\_gene: if True, use custom gene text file. File should contain 4 columns separated by whitespace: chromosome number, gene start, gene end, gene name. Chromosomes X,Y and mitochondrial (M) should be coded as 23, 24 and 25 respectively. If False, will use default genes on file.

genes2text: if True, will save text file of genes found

genes2screen: if True, will print genes found to screen

display\_chr: if True, will display and save chromosome number with genes

genes\_filename: name for .npy and .txt (if requested) file, where gene data is saved.

file\_folder: folder from which to save and load files. Defaults to current directory

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```
genefind_ncbi(snplist=[123434,12343557,2342342],bp_range=100000, data_verbose=True, gene2screen=True, file_folder="current", genefile="geneDF.p")
```

genefind\_ncbi: function for getting genes in the vicinity of a list of snps, such as those generated by evo\_triangularator, using ncbi gene databases

parameters:

snplist: numpy array or list of snp numbers to find nearby genes for

bp\_range: range of base pairs on either side of snp in which to search for genes

data\_verbose: if True, list of genes for each snp will be a list of dictionaries of various additional gene data:

gene name, chromosome, description, aliases, gene start position, gene stop position, summary

keyed as, respectively:

'Name', 'Chr', 'Description', 'Alias', 'Start', 'Stop', 'Summary'

gene2screen: if True, will print summary of genes found to screen

file\_folder: directory in which to save gene dataframes

genefile: file name to save dataframe

returns:

with data\_verbose == True: a list of two dataFrames, first one with simple lists of genes as final entry for each snp, second with lists of dictionaries with gene information for each snp (as described above)

with data\_verbose == False: just returns simple data frame

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```
evo_tri.hit_plotter2D(backend="TkAgg", granularity=10,
fst_file='fst_data1.npz', pops=['CEU','YRI','GIH'],
plot_snps=True, plot_genes=True,
pop12lim=">=X", pop13lim=">=.45", pop23lim="<=.05",
loc_data='pop_data1', bp_range=100000, file_folder="current")
```

hit\_plotter2D: function to plot number of hits (genes and/or snps) that the evolutionary triangulator finds against a changing Fst on one of the population-pair axes

parameters:

- backend: enter string to select matplotlib backend for generating figure
- granularity: how many points to plot between an Fst of 0 and 1
- fst\_file: Fst data to perform evolutionary triangulation data on. Should be a name of dictionary containing Fst data for pairings of three populations (such as Fst\_out from evo\_tri\_data() )
- pops: list of population abbreviations for population 1, population 2, and population 3, in that order
- pop12lim,pop13lim,pop23lim: Fst threshold (string with operator [>,>,>=,<=] followed by Fst between 0 and 1) for pop1-pop2 Fsts, pop1-pop3 Fsts, and pop2-pop3 Fsts respectively  
One and only one of these thresholds should be an operator followed by character X. This will be the axis along which the value changes.
- loc\_data: if plotting gene hits, provide a string with .npy or .npz file name (without extension), with file containing SNP location data, in format of pop\_out from evo\_tri\_data() .
- bp\_range: if plotting gene hits, provide an integer specifying how many bases from a SNP a gene must be to be considered a hit.
- file\_folder: folder from which to save and load files. Default to current directory

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```
evo_tri.hit_plotter3D(backend="TkAgg", granularity=10,
fst_file='fst_data1.npz', pops=['CEU','YRI','GIH'],
plot_snps=True, plot_genes=True,
pop12lim=">=X", pop13lim=">=Y", pop23lim="<=.05",
gene_maxZ=100, snp_maxZ=300,
loc_data='pop_data1', bp_range=100000, file_folder="current")
```

hit\_plotter3D: function to plot number of hits (genes and/or snps) that the evolutionary triangulator finds against a changing Fst on two of the population-pair axes. This function can take a long time to run, depending on granularity.

parameters:

- backend: enter string to select matplotlib backend for generating figure
- granularity: how many points to plot between an Fst of 0 and 1
- fst\_file: Fst data to perform evolutionary triangulation data on. Should be a name of dictionary containing Fst data for pairings of three populations (such as Fst\_out from evo\_tri\_data() )
- pops: list of population abbreviations for population 1, population 2, and population 3, in that order
- pop12lim,pop13lim,pop23lim: Fst threshold (string with operator [>,>,>=,<=]

followed by Fst between 0 and 1) for pop1-pop2 Fsts, pop1-pop3 Fsts, and pop2-pop3 Fsts respectively. One of these thresholds should be an operator followed by character X. This will be the first axis along which the value Changes. Another of these thresholds should be an operator followed by character Y. This will be the second axis along which the value changes.

loc\_data: if plotting gene hits, provide a string with .npz file name (without extension), with file containing SNP location data, in format of pop\_out from evo\_tri\_data().

bp\_range: if plotting gene hits, provide an integer specifying how many bases from a SNP a gene must be to be considered a hit.

gene\_maxZ: max of z-axis range for number of genes

snp\_maxZ: max of z-axis range for number of genes

file\_folder: folder from which to save and load files. Defaults to current directory.